



SLC25A15 gene

solute carrier family 25 member 15

Normal Function

The *SLC25A15* gene provides instructions for making a protein called a mitochondrial ornithine transporter. This transporter protein is needed for the urea cycle, a series of reactions that occurs in liver cells. The urea cycle processes excess nitrogen, generated when protein is used by the body, into a compound called urea that is excreted by the kidneys. Excreting the excess nitrogen prevents it from accumulating in the form of ammonia, which is toxic, especially to the nervous system.

The mitochondrial ornithine transporter protein moves a molecule called ornithine within the mitochondria (the energy-producing centers in cells). Specifically, this protein transports ornithine across the inner membrane of mitochondria to the region called the mitochondrial matrix, where it participates in the urea cycle.

Health Conditions Related to Genetic Changes

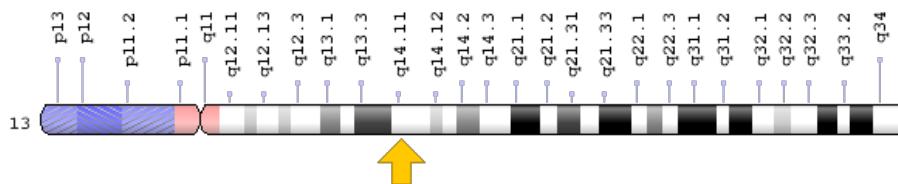
ornithine translocase deficiency

Approximately 17 mutations in the *SLC25A15* gene have been identified in individuals affected by ornithine translocase deficiency. The mutations result in a mitochondrial ornithine transporter that is unstable, the wrong shape, or otherwise lacking the ability to bring ornithine to the mitochondrial matrix. This failure of ornithine transport causes an interruption of the urea cycle and the accumulation of ammonia, resulting in the signs and symptoms of ornithine translocase deficiency.

Chromosomal Location

Cytogenetic Location: 13q14.11, which is the long (q) arm of chromosome 13 at position 14.11

Molecular Location: base pairs 40,789,411 to 40,812,460 on chromosome 13 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- D13S327
- HHH
- ORC1
- ornithine transporter 1
- ORNT1
- ORNT1_HUMAN
- OTTHUMP00000042249
- solute carrier family 25 (mitochondrial carrier; ornithine transporter) member 15

Additional Information & Resources

GeneReviews

- Hyperornithinemia-Hyperammonemia-Homocitrullinuria Syndrome
<https://www.ncbi.nlm.nih.gov/books/NBK97260>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28SLC25A15%5BTIAB%5D%29+OR+%28solute+carrier+family+25+++member+15%5BTIAB%5D%29%29+OR+R+%28%28HHH%5BTIAB%5D%29+OR+%28ORNT1%5BTIAB%5D%29+OR+%28D13S327%5BTIAB%5D%29+OR+%28OTTHUMP00000042249%5BTIAB%5D%29+OR+%28ornithine+transporter+1%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+2160+days%22%5Bdp%5D>

OMIM

- SOLUTE CARRIER FAMILY 25 (MITOCHONDRIAL CARRIER, ORNITHINE TRANSPORTER), MEMBER 15
<http://omim.org/entry/603861>

Research Resources

- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=SLC25A15%5Bgene%5D>
- HGNC Gene Family: Solute carriers
<http://www.genenames.org/cgi-bin/genefamilies/set/752>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=10985
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/10166>
- UniProt
<http://www.uniprot.org/uniprot/Q9Y619>

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